THE EUROPEAN SOCIETY OF HUMAN GENETICS
NEWSLETTER
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Report from the President

by Bill Newman,
President of the ESHG 2024-2025

Dear ESHG members,

Well, another year has flown by. For many, this has been an extremely difficult one, with conflicts raging across the world, economic instability, repeated reports of atrocities and a lack of kindness and generosity at the highest levels. We have seen attacks on scientists, doctors and the funding of important genomics research in a way that previously was hardly imaginable.

Despite this, this has been a year where there have been remarkable advances in genetics - a true realisation of the importance of the non-coding genome in neurodevelopmental and other rare disorders; new gene therapies have been adopted into clinical practice; large scale newborn screening projects have been initiated, and further fundamental insights have been made in genomic medicine. To be part of a community that is making discoveries and using science for broad benefit is invigorating and a source of light in darkened times. We have reached out to the American Society of Human Genetics and colleagues across other international genetics societies to see how we can most effectively work together to ensure that our ESHG voice drives policy and represents our community in protecting and enhancing genomic medicine for patient and public benefit. We are hoping that the International Federation Human Genetics Societies' meeting will take place in Guadalajara, Mexico in February 2027 and more details will be available soon.

Within the ESHG, there has been several new initiatives building upon the strong foundations lay down over previous years. I am amazed at the energy and commitment made by so many members to the work of the ESHG through the various committees, the organisation of educational courses, public policy and ethics work and the hosting of such a wonderful conference. The meeting in Milan will be the largest in terms of submitted abstracts that we have ever had and shows the rude health that human genetics experiences across Europe. This year has seen the start of the ESHG webinar series to complement the annual conference by providing a monthly opportunity to hear about world class genomic science. For details, please see the website https://www.eshg.org/webinarseries. We are always keen to have suggestions for future speakers.

Please take the opportunity to get involved in the Societies' activities. In particular, the ESHG-Y committee has brought a new perspective and energy to our organisation. Look out for information about our educational courses, engage with the webinars and enjoy the opportunity to meet colleagues and make new friends at the meeting in Milan. Every best wish to my successor as President of the ESHG, Professor Martina Cornel. It has been my privilege to be in the role for the past year working with the wonderful team at the Vienna Medical Academy and all of the Executive, Board and members.

Report from the Scientific Programme Committee

by Alexandre Hoischen & Alexandre Reymond, Co-Chairs of the SPC

We are Alexandre Reymond and Alexander Hoischen, joint co-chairs of the ESHG Scientific Program Committee (SPC). We are privileged that our SPC with >33 expert members represent the breadth of our discipline. They passionately aim to create the best possible program for our annual conference. Berlin ESHG 2024 broke several records: 6,544 participants human geneticists (5,490 registrants from 96 countries and 1,054 exhibitors), including 1,023 participants online.

We hope to see you even more numerous in Milan this year. Once again and for the fourth time, we will offer a fully hybrid conference, allowing geneticists who cannot attend in person to participate. We are pleased to propose an extremely attractive flat fees of just €100 (€50 for students) and even lower fees for participants outside EU. With this we support ESHGs mission to continue outreach and education to our global community.

For Milan's ESHG2025, we have put together a very exciting program of 3 award lectures, 39 sessions (29 symposia, 9 educational sessions and 1 plenary) totaling 111 invited speakers, 19 workshops for a more hands-on experience and 5 Get2Gether sessions to promote networking and professional connections.

We selected 210 abstracts for platform presentations and 48 for best poster flash-talk presentations from a total of another record-breaking 3,477 submitted abstracts (3,415 regular and 62 late-breaking abstracts)! Facing an ever-growing number of abstract submissions, we changed the conference setup to accommodate more concurrent session and offer platform presentations to >6% of submitted abstracts. This means that Milan 2025 will start with a full program Saturday morning. Many other high scoring abstracts will be presented as posters, again in its successful style as hybrid (top 66%) or e-posters. We also allowed a higher number of 48 of highly scoring posters the opportunity for poster flash-talks – a format that receives increasing attention in our dedicated 'sequencing square' within the exhibition hall. We are particularly thrilled that ESHG remains a meeting that offers a stage to many junior talents: Of all abstract selected talks 47% were submitted by Early Career Award (ECA) candidates. Participation of early career scientists is further supported by the >246 fellowships offered by ESHG (including ESHG conference fellowships, national fellowships as well as fellowships dedicated to Ukrainian scientists).

In these challenging times, we are excited to offer a very diverse program – with diverse topics that make our field so attractive, but also to attract broad and diverse participation. We are therefore thrilled that we can welcome abstracts from 83 countries. We are proud, that about half of all invited speakers are female. Like in Berlin, we have organized to have childcare service available to all participants and for the first time a "carer award" to invited speakers or committee members. To further increase the quality of our conference we will start offering in Milan text captions with our on demand videos to help non-native speakers and people with auditory challenges. We hope you will enjoy ESHG 2025 Milan as much as we will. We cannot wait to meet colleagues and friends from all over the world and engage, network and learn from each other.

We are looking forward to hosting you in Milan. Welcome to Italy – benvenuti a Milano!

Ciao,

Alex & Alex

www.eshg.org/spc

Report from the Policy and Ethics Committee

by Francesca Forzano, Chair of the PEC

The Policy and Ethics Committee (PEC) is an interdisciplinary Committee of the ESHG with the mission to contribute to the responsible translation of advancements in human genetics, by identifying and discussing the ethical, social and policy issues related to human genetics and its application in

research, clinical practice and laboratory genetic services.

It addresses these issues and provides guidance through background documents, policy statements, recommendations or other publications, and participating in the public and professional debate around these issues. The Committee collaborates with a range of other societies and organisations such as ELPAG, ESHRE, GA4GH and ASHG-Social Issues Committee.

Since 2023, the Committee took on an additional role, to provide ELSI support to the EJHG board in selected cases. An updated Terms of Reference has been produced this year and will be published soon on the PEC webpage.

In 2024-2025, the PEC has been active on the following topics:
A new, interactive course has been created by PEC and has launched in October 2024: Covering The Gaps - The Course You Didn't Know You Needed. The pilot version has been very successful, and a new edition will be offered on November 2025. The course will be run virtually on 2 days and will be particularly addressed to young scientists and clinicians. The course aims to cover topics that are important for your genetics research and practice but often neglected or taken for granted. It will cover issues of quality, regulation, ethics and justice in diagnostics and research. We will look at data sharing and biobanks, at returning results to patients and reporting research.
The document on Cascade Counselling and Testing, a strategy still underexploited across Europe, has been approved by the Board and submitted for publication to the EJHG. Thank you to all the members who have reviewed the document!
The document on the GDPR and what does it mean in practice for our genetic community has been reframed as Points to consider and is currently at the stage of PEC review for final approval. We plan to submit the manuscript for peer-reviewed publication on June 2025.
We are still finalizing the document on Misuse of genetic testing for discrimination purposes, we aim to complete it over summer 2025.

We ran a Survey on Opportunistic genomic screening, to ascertain the impact of the ESHG recommendations on this practice in European countries – thank you to all the ESHG members who have answered! We will present the results during the conference in Milan – check out the concurrent sessions!

On January 2025, we have run a workshop on Rare Diseases, Genomics and Justice at the Brocher Foundation. This was a collaboration between PEC – leader Angus Clarke - and a research team led by Ramona Moldovan funded by MRC UK. The workshop has been very successful, and all the participants provided excellent feedback. We are extremely grateful to all our distinguished invited speakers, who have provided excellent contributions. The outcome of the workshop will be published on a special issue of the Journal of Community Genetics, including a meeting summary and the key recommendations.

Polygenic risk scores in pre-implantation genetic testing. PEC members continue to be involved in discussions around this topic, and have contributed to workshops, webinars, seminars and conferences.
The members of PEC are working together with the EuroGentest Committee and the International Society for Prenatal Diagnosis to draft Recommendations on reporting genetic tests in prenatal diagnosis. The draft is in advanced stage and will likely be available for members' comments before the end of 2025.

A document on AI in genomics, in collaboration with Global Alliance for Genomics and Health (GA4GH), remains on our agenda for 2025. Another major topic on our radar is the interplay between Genetic testing and insurances. We are considering running a survey across membership in 2025 to understand what is currently happening across Europe in relation to use of genetic testing for modulating the insurance premium, access to genetic tests results from insurances, and offer of genetic tests from insurances.

ESHG members having suggestions for new topics to work on, or interested in working on a specific topic as a collaborator are encouraged to contact the PEC via the Chair. ESHG members interested in becoming a PEC member are requested to send their CV and a letter of interest stating their expertise and motivation for joining to the Chair.

Members of the PEC in 2024/2025 were: Angus Clarke, Christophe Cordier, Guido de Wert, Francesca Forzano (Chair), Sabine Hentze, Heidi Howard, Milan Macek, Béla Melegh, Alvaro Mendes, Yves Moreau, Markus Perola, Inga Prokopenko, Emmanuelle Rial-Sebbag, Rosaline Favresse (EURORDIS), Vigdis Stefánsdottir, Fiona Ulph and Carla van El (Secretary-general). Observers: Olga Antonova, Elena Avram, Yalda Jamshidi. ESHG-Y representative: Rhys Dore.

www.eshg.org/pec

Report from the Education Committee (EduComm) 2024-2025

by Inga Prokopenko, Chair of the EduComm

The ESHG is committed to advancing the excellence and understanding of Human and Medical Genetics across Europe.

Over the past year, the Education Committee has experienced a period of both productivity and enthusiasm, as we have made significant progress on multiple initiatives. We are pleased to welcome Julia Baptista, Hans Hennies, Eka Kvaratskhelia, Alisdair McNeill to our committee, who will focus on online courses, relationships with European Research Networks, mentorship programmes, online/digital resources on genomic testing.
We extend our heartfelt gratitude to Patricia Calapod, Johan den Dunnen, Vita Dolzan, Sally Ann Lynch, James O'Byrne, Raquel Silva Celia Soares, who have served on the committee since 2021.

ESHG Courses
Our ongoing commitment includes a diverse portfolio of in-person and virtual educational courses. For the latest list of courses and their details, visit https://www.eshg.org/courses. We report on new successful courses "**Introduction to large-scale genetic, epigenomic and transcriptomic data analysis**" (delivered in French), organised by Nabila Bouatia-Naji in Marrakech, Morocco and online course "**Covering The Gaps - The Course You Didn't Know You Needed**", led by Francesca Forzano and Angus Clarke. We are excited to mention a new edition of the hybrid "**Precision Genomic Medicine Workshop 2025**", led by James O'Byrne and developed in partnership with the Canadian College of Medical Genetics (CCMG) and the Human Genetics Society of Australasia (HGSA). We are looking forward to a new **Pharmacogenomics Course**, led by Vita Dolzan in Portorož, Slovenia. The committee welcomes suggestions from the membership to broaden the scope of our course offerings. For the supported courses, ESHG provides fellowships to young scientists and clinicians from economically disadvantaged countries to attend these programs.

The free **App-based courses, delivered by X-Peer** and launched in 2023, have attracted considerable attention. Over the past year, the channel has seen approximately 441 monthly users, 3,236 views, 76 completed tests, and 55 new diplomas awarded. The three modules cover key topics: diagnostic approaches to genetic syndromes and inherited metabolic disorders; developmental and epileptic encephalopathies and brain malformations; and movement disorders, neurodegenerative diseases, and neurocutaneous syndromes. To enrol in the course, please register for free at https://web.xpeer.app/web/en/courses/340?channelId=46. CME points are available for completing this program. We encourage you to explore this valuable resource and contribute suggestions for additional app-based courses. We are working on several new courses to be announced soon.

EuroGems update - led by Ed Tobias (Glasgow)
The ESHG educational website, https://www.eurogems.org, continues to receive an increasing number of visits, with users from 145 countries worldwide now accessing the site. We are delighted by this growing engagement and by the increased global accessibility resulting from offering the resource in four languages. Particularly noteworthy has been the growing number of visits to the professionally translated Spanish, Portuguese and French pages from numerous countries, within and outside Europe. Alongside the growing number of users, educators from many countries including Canada, Japan, Singapore, India, Egypt, South Africa, Argentina and Mexico have praised its usefulness. In addition, its web pages are now being used by the members of HUGO-International. Ed continues to enhance the website and to develop the associated **free educational genomics terminology apps**, available at https://www.genomicsapps.org. Comments and suggestions are welcome at edward.tobias@glasgow.ac.uk.

International Links – co-led by Edith Coonen, Liz Loehrer, Carmen Navas and Ed Tobias
With the focus on promoting genetic education, we expanded our connections, engaging with the Indian, Irish, Georgian, and Romanian national societies. We are connected with genomics educators providing clinical genetic training in Tanzania. We are developing a summary of meetings with stakeholders on the future needs for genomic education in Europe.

Podcast Program - curated by Sofia Douzgou Houge and in collaboration with the Whitworth group
ESHG, in collaboration with the Whitworth group

and the ESHG-Young Committee, has delivered Series 3 of Genetic Sounds, available at https://www.eshg.org/geneticsounds. The last two episodes were dedicated to Pharmacogenetics and Language: talking about genetics. This monthly podcast (released six times a year) delves into various genetic topics, now that genomics has taken centre stage in medicine. Using a conversational approach and featuring esteemed guest speakers, the podcast covers a wide range of subjects related to genetics and genomics. Since its inception, the podcast has consistently garnered high engagement across all continents. All episodes are accessible on multiple platforms, including Buzzsprout, Apple, and Spotify. Additionally, there will be a live podcast event at the ESHG conference in Glasgow, so be sure to watch out for it on the program. We believe this podcast is an effective way to engage with both professionals and the general public, and we welcome ideas for future topics. Furthermore, we have established a partnership with the EURORDIS Rare on Air podcast, available at https://www.eurordis.org/rare-on-air/ .

International Mentorship and Observership Schemes

We have had lots of interest in the new ESHG mentorship and observership schemes.

The mentorship scheme https://www.eshg.org/education/eshg-mentorship-programme
Offers up to five awards (up to €1500) for early career geneticists to visit another centre and attend the ESHG conference. The award will establish a long-term career supportive relationship.
One of the mentorship awards from 2024 onwards is ring-fenced for genetic counsellors to apply for and named in honour of Professor Lauren Kerzin-Storrar who encouraged trainees to travel to broaden their experience and knowledge and is a dearly remembered colleague. We encourage genetic counsellors to apply for the Mentorship award.

Awardees of the ESHG Mentorship scheme for 2025 are:

- **Tea Mladenic** Croatia to Germany
- **Daniela Oliveira** Portugal to Sweden
- **Lein Dofash** Australia (Palestine) to UK
- **Melda Erdogdu** Turkey to Sweden
- **Nesibe Saliha Bulut** Turkey to Austria

The observership scheme https://www.eshg.org/education/eshg-observership-programme offers funding (up to €2,000) to support 4 weeks for a young geneticist to visit another centre.

Awardees of the ESHG Observerships scheme for 2025 are:

- **Juliana Miranda Cerqueira** Finland to UK
- **Vanessa Sousa** Portugal to Belgium
- **Purvi Majethia** India to UK
- **Luiza Lorena Pires Ramos** Belgium to Sweden
- **Sílvia Pires** Portugal to Germany

The next round of applications for these two programs will be live in August with a closing date in November 2025.

DNA Day Essay and Video contest – led by Christophe Cordier

DNA Day, celebrated on April 25, commemorates the groundbreaking discovery of DNA's structure in 1953 and recognises the profound impact of genetics on science and medicine. For the 17th year, the ESHG has sponsored a European DNA Day essay and video contest for high school students, encouraging exploration of key issues in genetics.

This year's theme highlighted the fact that «For many years in the past century, geneticists focussed on isolated populations to discover genetic traits, including lactose intolerance or sickle cell anaemia. Recent advancements in genetic and genomic technologies have enabled the conduction of large-scale studies in human genetics, such as those by the UK Biobank, Estonian Biobank, and FinnGen. However, still do not encompass the full diversity of world populations». Students were asked to discuss why understanding genetic diversity is vital for health and disease, using specific genes or DNA variants as examples. The contest aims to raise awareness about the importance of diversity in genetics and inspire young people to engage with its role in advancing equitable healthcare and personalised medicine. Winners will be announced at the ESHG conference in Milan.

If you have any ideas/comments regarding the work of EduComm please contact me at i.prokopenko@surrey.ac.uk .

www.eshg.org/educom

Annual Report ESHG-Euro-Gentest Committee 2024

by Gert Matthijs, Chair of the EuroGentest Committee

The ESHG-EuroGentest committee focuses on quality aspects of genetic services, preparing professional guidelines related to genetic diagnostics, providing information, and representing the society's interests at international forums related to genetic services. The EuroGentest committee also prepares ESHG's policy and training activities regarding the In Vitro Diagnostic Regulation (IVDR).

The 2024-2025 EuroGentest committee members are Christi van Asperen, Isabel Maria Carreira, Christophe Cordier, Donna Darmanin, Els Dequeker, Jenni Fairley, Weronika Gutowska-Ding, Thomas Liehr, Milan Macek Jr, Aleš Maver, Joana Barbosa Melo, Michael Morris, Katrin Õunap, Simon Patton, Rebecka Pestoff, Christine Vianey-Saban and Nicola Wolstenholme. We gratefully acknowledge the time and effort they spent to voluntarily contribute to ESHG-EuroGentest achievements and output over the last year.

On April 1st, EuroGentest organized the online seminar «**Navigating the future of genetics**» with updates on quality assurance, regulations and professional standards in clinical and laboratory practices. The contributions included a summary of EuroGentest-ESHG's feedback in the context of the European Commission's targeted IVDR evaluation and an example of an IVDR trajectory for comprehensive cancer testing, an introduction to rapid exome sequencing and the genetic counsellor's and patient's perspective thereon, a quick guide to external quality assessment and an overview of professional recognition in the genetics arena. The recordings of the presentations are freely available (registration required; see https://www.eshg.org/courses/eshg-eurogentest-webinar#c6428).

IVDR-related activities: the ESHG-EuroGentest Task Force on IVDR (Els Dequeker, Milan Macek Jr, Gunnar Houge, Thomas Liehr, Gert Matthijs, Joris Vermeesch)

The IVD Regulation poses a significant challenge for genetic diagnostic laboratories, particularly regarding the use of in-house developed tests when no CE-IVD kit is commercially available for the same diagnostic purpose. The joint ESHG-EuroGentest Task Force on IVDR aims to inform diagnostic laboratories and raise awareness within the genetics community and towards regulatory authorities about key concerns related to the implementation of the IVDR. Leading this effort is Els Dequeker, who was previously appointed by the ESHG to represent the society in the BioMed Alliance, a coalition of European medical societies engaged in lobbying at the European level. Through this role, she became a member of the European Commission's Task Force Medical Device Coordination Group (MDCG) on In Vitro Diagnostics and IVDR Orphan Diagnostics.

Over the past year, efforts have continued to highlight concerns regarding the loss of innovation and the availability of IVD devices for orphan diagnostics. A dedicated MDCG working group on IVDR orphan devices was established and has already convened several times. Through the ESHG – BioMed Alliance, we had the opportunity to provide examples of orphan devices that currently remain unaddressed in the IVDR and its associated MDCG documents.

In response to the call for evidence on the IVDR, a survey was distributed to all ESHG members. We sincerely thank all our colleagues who have responded to the survey!

Thanks to the strong response, we were able to submit a well-documented report in March 2025, reinforcing seven key points with data from the survey to highlight the challenges genetic services face under the current IVDR framework.

The main challenges identified include:

1. Ensuring harmonization in monitoring IVD device safety within healthcare institutions.
2. Clarifying the distinction between CE-IVD devices and in-house IVD devices.
3. Addressing the shortage of CE-IVD devices in genetic testing while maintaining space for innovation.
4. Assessing the requirement to use CE-IVD equipment.
5. Evaluating the added value of the IVDR declaration of conformity.
6. Securing the use of in-house IVDs for orphan diagnostics.
7. Defining the legal entity status of healthcare institutions, including genetic laboratories.

(For the entire document, see https://ec.europa.eu/info/law/better-regulation/have-your-say/initiatives/14155-EU-rules-on-medical-devices-and-in-vitro-diagnostics-targeted-evaluation/F3530219_en).

We also embarked on the following projects:

Harmonization in genetic counselling (Rebecka Pestoff, Christi van Asperen, Christophe Cordier, Donna Darmanin, Katrin Õunap)
This is a challenging issue. Aspects of quality in clinical practice are more difficult to evaluate and impose than in the laboratory. However, it is essential to provide quality assurance and patient safety within rapid developments in the field. A summary of the current published state-of-the art regarding the harmonization of the profession of genetic counsellor in Europe is underway. It is an important first step in describing the current evidence base and in identifying issues and potential solutions. In close collaboration with the European Board of Medical Genetics (EBMG), we intend to report regarding the current situation in Europe.

Poor performance in EQA (Weronika Gutowska-Ding, Christine Saban, Els Dequeker, Jenni Fairley, Nicola Wolstenholme)
The Joint Committee on Poor Performance was established to address challenges related to poor performance in External Quality Assessment (EQA) across Europe. This committee brings together representatives from key EQA providers, including Equalis, Instand, ERNDIM, UK NEQAS for haematology, GenQA, and EMQN, ensuring a broad and collaborative approach to improving laboratory performance standards.
The committee's primary goal is to collect, compare, and harmonise the definition of poor performance among different EQA providers. Currently, discrepancies in assessment criteria, thresholds, and corrective action protocols exist across organisations, leading to inconsistencies in how underperformance is identified and addressed and confusion amongst the participants. By working towards a unified definition, the committee aims to establish a standardised, transparent, and fair assessment framework applicable across all participating EQA schemes. This will enhance clarity for laboratories, promote comparability of performance data, and ultimately lead to higher testing standards across Europe.
Beyond defining poor performance, the committee will focus on developing effective intervention strategies for laboratories that demonstrate consistent underperformance in EQA schemes. The challenge lies in determining how to respond to and support these laboratories in a way that ensures improvement while maintaining patient safety and diagnostic accuracy.
Given that the ESHG serves as a professional body with links to national genetics societies, it provides an ideal platform for implementing these measures in a coordinated and structured manner. By leveraging the influence and expertise of the ESHG, the committee can facilitate policy discussions, encourage best practices, and promote a culture of continuous quality improvement in genetic testing laboratories across Europe. Through these combined efforts, the Joint Committee on Poor Performance aims to strengthen the overall reliability and quality of genetic diagnostic services, ensuring that patients receive accurate and high-standard results regardless of the laboratory performing the test.
If any additional representatives from EQA providers are interested in joining our Committee, please reach out to Weronika at eurogentest@eshg.org.

Update of FMR1 guidelines (Weronika Gutowska-Ding)
Best practice guidelines reflect what peers think about good practice in diagnostics and quality management. Professional guidelines are useful as a basis for the accreditation of laboratories and may be used as a reference to good practice and standard-of-care in juridical disputes. A working group has been created to update the guidelines for molecular testing and genetic reporting of Fragile X syndrome (FXS). The available document dates back to 2014, and the technique that is mostly used to test the FMR1 expansion has now changed to be based on triplet repeat PCR.

Reporting in prenatal testing (Michael Morris, Luca Lovrečić, Lina Basel)
During 2024, the Policy and Ethics Committee (PEC) of ESHG identified the need for standardisation and guidance on the delivery of diagnostic fetal sequencing. In parallel, the International Society for Prenatal Diagnosis (ISPD) aimed to draft similar guidance, therefore under the leadership of Prof. Sandi Deans, GenQA, both professional bodies have collaborated to create a global authorship to produce a position statement with high-level recommendations for safe clinical practice. The manuscript is nearing completion, and we hope it will be published simultaneously in the European Journal of Human Genetics and Prenatal Diagnosis.

If you are interested in joining the activities of EuroGentest, please let us know: eurogentest@eshg.org. More information and useful resources including information leaflets for patients and families are available at **www.eshg.org/egt**

Report from the ESHG Young Committee

by Mridul Johari, Chair of ESHG-Y on behalf of all Committee Members

The European Society of Human Genetics-Young Committee (ESHG-Young) continues its dedicated mission of nurturing and empowering emerging geneticists throughout Europe. During 2024-25, ESHG-Young has served as a catalyst for educational advancement, community building, and professional development, creating pathways for the rising generation of innovators in human genetics.

2025 brings significant changes to the ESHG-Young board. Three new members will join the board with the election results being announced during our Get-2Gether session on Saturday, May 24th at 12:15. The elected members will replace Mridul Johari, Delia Sabau, and Raquel Silva, who have served since June 2021. The newly created chair-elect position allows for smoother transitions, with Silvia Kalantari appointed to serve as the Chair from June 2025. Nelson Martins joined as an associate member in January 2025, supporting the social media team, while Juliana Miranda Cerqueira's consultant role was extended to June 2026 for her valuable work in the Scientific Program Committee.

Mridul Johari (Chair) and Juliana Miranda Cerqueira (Consultant) have maintained their representation of ESHG-Young, making significant contributions to the ESHG Scientific Program Committee (SPC). Their involvement has been instrumental in crafting a stimulating and enlightening program for the ESHG 2025 conference. The year 2025 will mark Mridul's final year representing the ESHG-Young in the ESHG-SPC. Rita Barbosa-Matos is set to succeed him while Juliana continues her role until 2026.

Ana Raquel Silva (Vice-Chair) and Silvia Kalantari (Chair-elect) have continued the historical collaboration with ERN-ITHACA, participating in the scientific committee for the 2024 and 2025 Eurodysmorpho conferences, and attending the board meeting of the society in Bucharest (December 2024). The next Eurodysmorpho will be held in Vilnius (17-24 Sept 2025) and will feature a new pre-conference session on risk calculation in clinical genetics with Dr. Emilia Bijlsma, completely dedicated to young geneticists in training.

Ana Raquel Silva and Rita Barbosa-Matos (Secretary) continued to strengthen ties with EduComm as ESHG-Young representatives. They reviewed and supported the applications of early-career geneticists through the EduComm 2025 Mentorship & Observership Fellowship Programs. In the near future, we hope to enhance our collaboration on the ESHG Podcast, selecting new hot topics to be featured in the next series.
The ESHG-Young SM team (Rita Barbosa-Matos, Magdalena Mroczek, and Juliana Miranda Cerqueira) has promoted content for young geneticists while supporting ESHG initiatives highlighting professional development opportunities. They manage our X, LinkedIn, and Bluesky accounts and publish the annual newsletter covering our activities and new member elections. Nelson Martins joined as an associate member focusing on the newsletter and visuals, while Rita and Juliana continue serving on the ESHG Social Media Committee.

Rhys Dore continues as the young representative on the Policy and Ethics Committee (PEC), attending biannual meetings at the ESHG Conference and in Geneva while contributing to proposals and document reviews. He plans to survey young geneticists about their policy and ethics educational needs to inform relevant PEC sessions. Meanwhile, Magdalena and Delia (Vice-Chair) remain ESHG-Young representatives on the European Journal of Human Genetics Editorial Board.

Magdalena Mroczek continues leading the African Society of Human Genetics (AfSHG) collaboration, attending the 15th AfSHG Conference and 1st Ugandan Society meeting in February 2025. A cell disease models webinar with JAX Laboratories is being organized for 2025.
Delia Sabau (Vice-Chair) has continued her role within the ASHG Membership Engagement Com-

mittee, participating in ASHG 2024 Annual Meeting in Denver, Colorado. Meanwhile, Mridul Johari has strengthened his position in the working committee of the Human Genetics Society of Australasia (Western Australia branch), further enhancing our cross-continental partnership initiatives.

Delia Sabau (Vice-Chair) has continued her role within the European Board of Medical Genetics (EBMG), organising together with Professor Dr. Jonathan Berg and Dr Laura Polsler the annual Zoom meeting (5th Dec 2024) for medical doctors, providing information and details about the European Certificate in Medical Genetics and Genomics . Last year's session provided candidates a perspective of a successful candidate, Dr. Sophie Uyttebroeck, who shared her experience of the examination.

Following its successful first edition, the Get2Gether session „Uniting European Young Initiatives in Human Genetics" returns on Saturday, May 24th at 12:15, featuring representatives from Albania, Denmark, France, Italy, Portugal, Romania, and Turkey. Organized by Silvia and Mathys Weber (Associate Member) with Mridul and Juliana from SPC, the discussion will explore national genetics training systems, career opportunities, and future directions. The team created an internal database of young geneticist societies across Europe, establishing a valuable resource for ongoing international collaboration within ESHG-Young.

After the success of the last two years, the ESHG-Young matchmaking event will be held in the timeslot **12.15-13.15 on Sunday 25th and Monday 26th** May at the ESHG-Young booth during the ESHG 2025 conference in Milan. The event is designed to connect trainees, mentors, and recruiters in a dynamic setting around tables subdivided by topic (i.e. companies, basic research, clinical research and a table dedicated to mentors). In order to make networking easier, ribbons will be used to identify participants.

ESHG-Young remains firmly committed to enhancing the visibility and education of early-career geneticists. With ambitions to broaden its impact and scope, ESHG-Young is focused on strengthening global partnerships and advocating for crucial advancements in both education and policy within the genetics field. We stand prepared to shepherd young geneticists toward an innovative and inclusive future in human

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Social Media Committee Annual report May 2025

by Aleena Mashtaq Stolworthy and James Fasham, Co-Chairs of the SMC

This year has seen significant political and social change reflected in changes in social media engagement and usage by the scientific community. The social media committee has positioned itself to enable ESHG to respond to these changes in a professional and considered but timely manner. Actions taken include the diversification of outputs to include Bluesky, a platform where an increasing the amount of scientific discourse is now concentrated.

The committee has also integrated Buffer and Canvas into its strategy. Buffer is a social media management tool what enables cross-posting, audience engagement, and performance tracking across X, Bluesky, Instagram, LinkedIn, and Facebook. Insights from Buffer's analytics, such as engagement rates (likes, comments, shares, reposts), follower growth, and high performing posts will help measure our reach and refine future content. Canvas enables the prompt generation of visuals.

The committee chairs hosted the 'How to use social media to communicate your science' workshop at ESHG 2024, with the primary goal of empowering researchers, clinicians, and counsellors to effectively use social platforms to build their online presence, connect with audiences, and highlight their scientific work through strategic engagement. This was well attended and prompted lively engagement and discussion.

Following this success we are planning an even more interactive session this year, entitled "Has social media broken scientific knowledge sharing?" This seeks to provide a forum for the scientific community to discuss the significant changes that have happened in the last year. There will also be a dedicated segment on how to counter misinformation in the age of viral

content. We have invited guest speakers with strong social media influence to share their insights, strategies, and personal experiences navigating this fast-evolving digital space. Their participation promises to elevate the workshop's impact, offering attendees new perspectives for online science communication. The committee has also supported other ESHG activities, with Aleena Mushtaq Stolworthy leading a social media workshop for the first edition of 'Covering The Gaps – The Course You Didn't Know You Needed' in October 2024. The committee has been invited once again to contribute, reflecting the growing demand for guidance in navigating the dynamic landscape of social media platforms.

Dr. James Fasham and Dr. Aleena Mushtaq

www.eshg.org/smc

Report from the Strategic Committee

by Borut Peterlin, Chair of the SC

In 2023, the European Society of Human Genetics (ESHG) established the Strategic Committee (SC) to shape the society's vision and affirm its position as a leading European and international hub in genomic medicine. As genomics increasingly becomes integral to medical practice amid rapid technological progress, we recognize the pivotal role that genetic factors play in human health. This recognition is enhancing the vital contributions of genetic professionals and steering the development of more personalized medicine and advanced therapeutic strategies, thereby transforming how healthcare is delivered and managed.

The SC's mission is to foster the translation of cutting-edge genomic science into clinical practice, enhancing diagnosis, prevention, treatment, and care for genetic and complex diseases.

Over the past year, our primary focus has been to develop the strategic direction for ESHG for 2025-2028. In addition to inputs from SC and ESHG Board members and individual experts, we conducted a comprehensive survey involving 386 geneticists from 52 countries worldwide. This survey highlighted key priorities such as integrating genomic medicine across medical specialties, developing clinical practice guidelines, influencing genomic healthcare policies, monitoring challenges in medical genetics across the EU, and developing applications for precision medicine.

Based on this feedback, we crafted our strategic approach around five main pillars: Integration of Genomic Medicine in Clinical Practice, Influencing health policies, Genomics for Prevention, Education, and Research.

Additionally, the SC is developing a position paper that will delineate the role of genetics and genetic professionals in future healthcare systems.

I am deeply grateful to all SC members and contributors for their insights and efforts in developing this strategic vision.

For any suggestions or comments regarding the Strategic Committee's work, please contact me at borut.peterlin@kclj.si.

www.eshg.org/sc

A review of activity in the EJHG 2024-2025

by Alisdair McNeill, Editor in Chief EJHG

In 2024, the European Journal of Human Genetics, the official journal of the European Society of Human Genetics, published a wide spectrum of genomics research in the form of original research, brief communications and review papers. This covered the spectrum from population genetics, rare disease gene discovery through to social science aspects. From 1st January 2024 to 31st December 2024 the European Journal of Human Genetics received 1,036 submissions (compared to 823 for the preceding 12

month period). Our acceptance rate is 19%. In 2024, there were well over 3.2 million downloads of articles published in the European Journal of Human Genetics and over 3000 altmetric mentions. Publications were submitted from a range of different countries - amongst accepted manuscripts the top 3 submitting regions were the United States of America, England and Germany. We continue to try and broaden our reach and welcome submissions of relevant research globally. Acceptance for publication in the European Journal of Human Genetics remains competitive and it remains a prestigious home for your research and review papers.

The 2023 impact factor of the European Journal of Human genetics is 3.7 (with a total of 11 583 citations). This was a significant fall from the previous impact factor of 5.2. This was disappointing, but reflects a general trend of increasing competition, with more journals in the field. There was a general fall in impact factors across comparator journals. According to the impact factor the European Journal of Human Genetics is ranked 53rd out of 191 Genetics and Heredity journals. The latest (2022) citescore of the European Journal of Human Genetics has risen to 9.1, and the journal is now ranked 11th out of 90 Genetics (Clinical) journals by Scopus. Our strategy to boost the impact factor of EJHG centres around improved social media promotion of papers and increasing the number of review papers we publish (since these are more highly cited). We would welcome submissions of relevant review papers and suggestions for review topics.

In 2024 we further developed our social media strategy to promote work published in the European Journal of Human Genetics. We appointed Dr Seda Zonuzi as our new social media editor. She is based at the journal editorial office at the University of Sheffield. The European Journal of Human Genetics X (Twitter) account has around 7 500 followers. This remains an important platform. We have also established Instagram and Linkdin accounts for EJHG. Across these platforms we will continue to promote EJHG papers by running online journal clubs and posting brief summaries of new papers as they are published online.

In the previous 12 months we have said farewell to colleagues who have left our editorial board (Angus Clarke, Fabiano de Oliveira Poswar, Stanislas Lyonnet, Jean-Louis Mandel and Orsetta Zuffardi) and welcomed several new members. New members include: Nadeem Qureshi, Amy Nisselle, Jeanne Amiel, Marco Savarese, Elena Semina, Linda Reis, George Wiggins, Michael DeGiorgio, Christian Dina, Megan Leask, Emma Wade, Tara Clancy, Claire Morgan, Jade Howard and Ashish Kapoor.

I will be at ESHG 2025 in Milan, please do let me have your thoughts on the journal, and ideas for its development, in person, if you wish. Once again, thank you to all the peer reviewers, editorial board members and section editors who make our journal function. A special mention goes to our editorial assistant Dr Shona Kirk.

www.eshg.org/ejhg

European Board of Medical Genetics - Report 2024

by Joana Barbosa de Melo, Clinical Laboratory Geneticist, President

The **European Board of Medical Genetics** (EBMG) is dedicated to establishing qualification and certification standards for professionals in the field of medical genetics. Its primary goal is to ensure excellence and high levels of competence in clinical genetics, laboratory genetics, and genetic counseling across Europe. By standardizing professional qualifications, the EBMG promotes the harmonization of training and certification across different European countries. Throughout this year, the EBMG has remained focused on strengthening its internal organization, including refining financial distribution strategies among its branches. Efforts have been directed at enhancing collaboration between the three branches, facilitated by regular monthly online meetings.

A significant area in progress has been the continued development of structured documentation on board positions and roles. These efforts aim to preserve institutional knowledge and ensure continuity when leadership transitions occur.

Additionally, the Board has addressed the growing role of bioinformaticians in medical genetics. Identifying the need for a formalized system of recogni-

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tion, the EBMG invited professionals from the field to collaborate on future initiatives that can start the establishment of a standardized framework for their certification.

The three Branches

The **Clinical Laboratory Geneticist Branch (CLG)** now has 400 registered professionals from approximately 40 countries, with 15 applications pending. The Board continues its dedicated efforts toward achieving European-level recognition for the profession through engagement with the European Commission.

The **Genetic Nurse & Genetic Counsellor Branch (GN&GC)** is currently completing assessments for first-time applicants; 12 new portfolios have been evaluated in the current cycle. A total of 140 genetic counsellors are currently registered across Europe. The branch will also be renewing registrations for professionals who were registered up to 5 years ago. Additionally, the branch is evaluating new MSc programs submitted from across Europe for EBMG accreditation; currently, there are 9 master's programs accredited by the board. Furthermore, the branch is developing new strategies to promote professional recognition at a European level.

The **Medical Geneticists (MG)** branch is, together with the UEMS section of Medical Genetics, running the ECMGG (European Certificate of Medical Genetics and Genomics). This exam is now fully appraised by CESMA, the Council for European Specialists Medical Assessment. Sixty-two candidates are planned to sit the exam this year (MCQ part on April 28th and oral part on June 16th). Also, 2024 was an important year for the branch because medical genetics was finally recognized as a specialty in Spain, in November 2024.

Outreach and Dissemination

To increase awareness of the EBMG's mission and activities, the Board was represented in the EuroGentest webinar on April 1st, 2025, where it presented the objectives, initiatives, and achievements of its three branches. More than 100 participants attended the webinar.

As part of its ongoing outreach efforts, the EBMG has also focused on updating its website and preparing the Get2Gether session at the 2025 European Society of Human Genetics (ESHG) conference in Milan: **Monday 26th May 2025 at 12:15 CEST**. This session will explore, with examples, how the formal recognition of the three genetic professions can benefit patients. Bioinformaticians are also strongly invited to participate in this event, fostering the development of their professional community within the EBMG framework.

With these initiatives, the EBMG continues to play a pivotal role in advancing medical genetics, ensuring that patients are met with professional excellence, and strengthening collaboration within our field. We deeply appreciate the efforts of all EBMG members and affiliates for their contributions. Their dedication is the foundation of the EBMG mission, fostering collaboration, mutual support, and respect among our professional branches, all committed to delivering high-quality genetic care.

www.ebmg.eu

Interview with Daniel MacArthur, ESHG Award Lecturer 2025

by Mary Rice, ESHG Press Officer

Professor Daniel MacArthur heads the Centre for Population Genomics, based in Sydney and Melbourne, Australia. He will be giving the ESHG Award Lecture on Tuesday 27 May at 14.15 hrs

Daniel MacArthur grew up in a medical family. "My father was a clinician and my mother a physiotherapist, so I grew up with conversations about the human body and how it works at a mechanistic level." But he didn't decide on a scientific career until much later. "I had always loved the process of distilling ideas into the written word, so I had assumed my career path would be something like journalism. It honestly wasn't until the week before I submitted my university preferences that I fully realised that I was far more interested in a path in science or medicine."

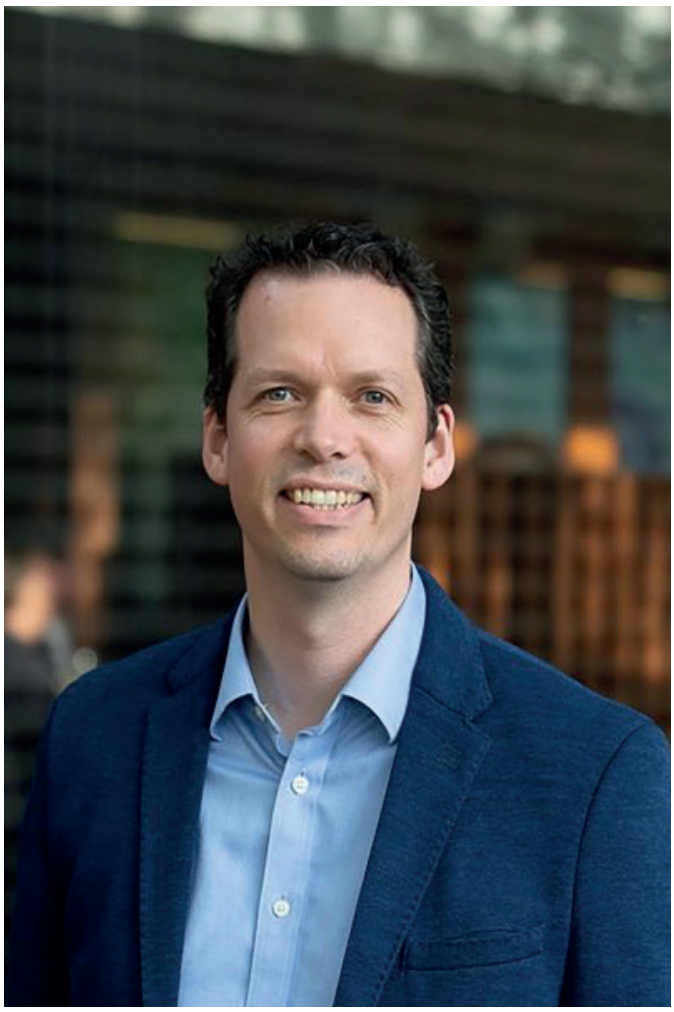
As a result, he did his undergraduate degree in biomedical science at the University of Sydney, Australia. Although this course was primarily designed as the first step for students planning to pursue a graduate medical course, he was still undecided about whether to go down the research or clinical path.

His first exposure to research made up his mind. "I did a summer student placement in the lab of Professor Kathryn North. I was working on a set of patients who had the class of muscle disorders known as muscular dystrophy, but who did not yet have a genetic diagnosis. During my placement I was able to find a particular protein that was missing in the muscle of two of the patients, and then sequence their DNA and find the causal mutations. Both patients had been waiting for a diagnosis for over a decade, and this hooked me completely on research: the intoxicating idea that, with a particular set of experiments, it was possible to come up with an answer that was not just interesting scientifically but could deeply change patients' lives."

To come up with something so fulfilling so early in a career is quite unusual, and MacArthur gives full credit to Kathryn North. "Giving an undergraduate student the chance to try something really important in such a short research project was remarkable," he notes. He went on to complete an Honours research year and then a PhD with North, who remains an important mentor. "So much of what I've learned about research, collaboration, and leadership has come from Kathryn," he says.

After a postdoc project at the Wellcome Trust Sanger Institute in the UK in 2012, MacArthur moved to the Broad Institute of MIT and Harvard, US, to build a new research team in a recently established department led by renowned statistical geneticist Mark Daly. Among other projects, MacArthur's team began working with exome sequencing to improve the diagnosis of patients with very rare genetic disorders. But they quickly realised that what they were missing was the existence of good reference datasets to give a picture of normal genetic variation in the general population. "At that time we had a resource that contained just over six thousand individuals, was pretty outdated in terms of data quality, and just wasn't large enough to allow accurate diagnoses."

However, he was surrounded by a group of scientists at the Broad Institute who were involved in generating exome sequencing on a massive scale. "They had already sequenced tens of thousands of people, mostly as part of large case control studies of the genetics of common complex disorders, like type 2 diabetes and coronary heart disease. And over the course of a few months I worked with leaders at the Broad – especially Mark Daly, David Altshuler, and Sek Kathiresan – on a really ambitious plan to collect all the human sequencing data being generated at the Institute,



and to aggregate it into a single, unified resource of genetic variation that we could share with the world."

That resource became the Exome Aggregation Consortium (ExAC), launched in 2014, which had an immediate impact on the diagnosis of rare disorders worldwide. It was renamed two years later as the Genome Aggregation Database (gnomAD), and has continued to grow over time, now encompassing exomes and genomes from nearly a million people. "This work was both extremely challenging and deeply rewarding because the sheer scale required people with lots of different backgrounds – data scientists, software engineers, computational biologists, project managers – to come together to solve fundamental problems in both genetics and computation, " he says. "But the impact has made it all worthwhile – millions of patients worldwide have now benefited from diagnostic tests that use the gnomAD resource."

MacArthur credits the success of gnomAD to being in the right place, at the right time, with the right people. "I was so lucky to have an incredible series of postdocs, computational biologists, and engineers in my group who did a lot of the heavy lifting, but we could

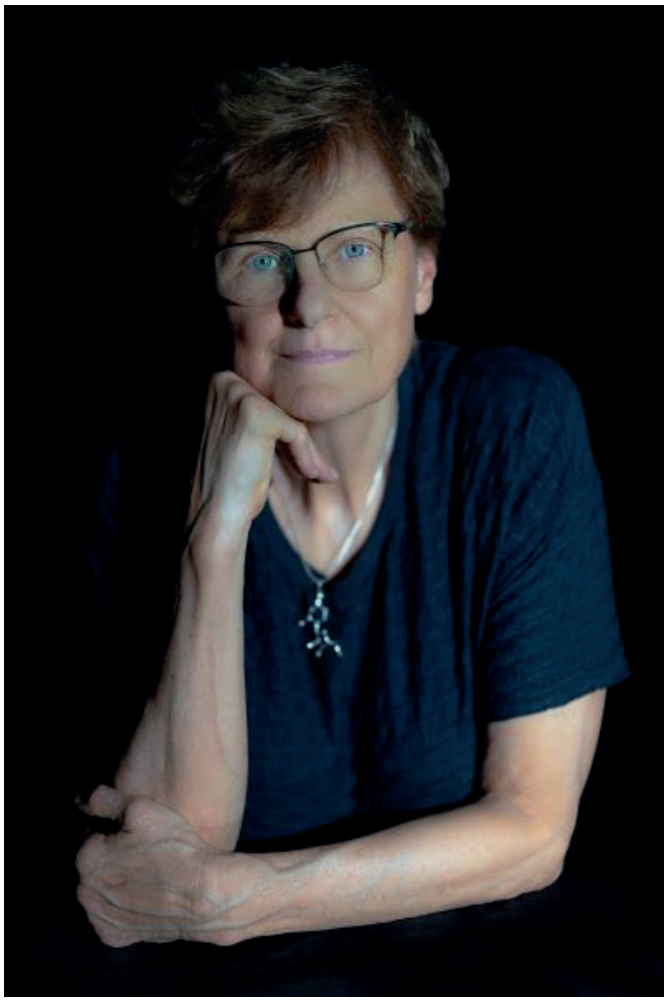
never have pulled it off without a much larger team including data scientists, engineers, project managers, and many other experts across the Broad Institute, and of course the many researchers and participants who donated their data to the programme."

He left the Broad to come back to Australia in early 2020, and is now Director of the Centre for Population Genomics (CPG), a joint initiative of the Garvan institute of Medical Research in Sydney and the Murdoch Children's Research Institute in Melbourne. The work of the CPG is focused on addressing challenges to the equitable, population-scale implementation of genomic medicine. The Centre's major programmes include OurDNA, which is currently recruiting 10,000 Australians of diverse ancestry and collecting DNA and live cells, and a rare disease programme that has already provided diagnoses to over 450 families affected by severe genetic conditions.

The work of gnomAD continues to thrive under the leadership of Heidi Rehm and Mark Daly, and MacArthur is still an active contributor to the program. "It's so common in academia for projects to die when a researcher moves on, and I'm really grateful that Heidi, Mark, and the broader team have not just kept gnomAD alive, but continued to massively grow and improve it."

MacArthur's time outside of work is largely focused on his family. "I have three young boys, and it's such a privilege to spend my time with them as they grow and develop into increasingly awesome human beings."

In his lecture, he will describe the origins of gnomAD and the power of open science, making data available early and as easily as possible, as well as the work he is currently leading in Australia. "I'll be covering the need to build genomic resources that are designed from the outset to lay the foundations for the next phase of genomic medicine, and to make sure its impact is equitable."



Interview with Katalin Karikó, Mendel Award Lecturer 2025

by Mary Rice, ESHG Press Officer

Katalin Karikó is Professor at University of Szeged, Hungary and Adjunct Professor at Perelman School of Medicine at the University of Pennsylvania, USA. She won the 2023 Nobel Prize in Physiology or Medicine and will be giving the Mendel Lecture on Tuesday 27 May at 13.30hrs.

Katalin Karikó remembers her first vaccination campaign quite clearly. "Students from the veterinary school came into our yard, and my sister and I handed over our chickens to them, one by one." And science lessons were all around her in her childhood in the Hungarian countryside – birds nesting, hatching, and migrating, soap-making, tackling potato pests, and learning about wildflowers and how they grow.

But it was in elementary school that she first had a formal introduction to science. "I joined a chemistry club. We constructed elements from playdough and then formed them into molecules using toothpicks." And the biology teacher encouraged their interest in the natural world by taking the class out for walks to observe such things as the mechanism that allows waterlilies to float.

You might think such a precocious interest in chemistry and biology led naturally to early excellence in the subjects, but that was not the case, she says. "I compensated with effort for what I lacked in natural ability. I worked and studied, and I got better. By third grade I was earning straight 5s." By eighth grade she was the best biology student in the town, then in the county, and then third in the whole country. After finishing high school, she started to study biology at the University of Szeged, where she is now a professor.

She performed her PhD and post doc studies at the Biological Research Centre of the Hungarian Academy of Sciences, also in Szeged, before leaving for a post doc position at Temple University, Philadelphia, USA, and the University of Health Science, Bethesda. In 1989 she was appointed Research Assistant Professor at the University of Pennsylvania, where she remained until 2013. After that she became vice president and later senior vice president at BioNTech, Mainz, Germany. Since 2021, in addition to her professorship at Szeged, she remained an Adjunct Professor at the University of Pennsylvania.

Her interest in the possibility of using RNA for medical purposes started at the Biological Research Centre, where she was researching interferon. "It was at a time when the molecular mechanism of interferons and their antiviral activity was just beginning to be understood. We were making so-called 2-5A RNA molecules in lab and testing out their antiviral effects." Experiments were going well when the funding was pulled, and she had to make the difficult decision to emigrate to the US. A letter to Dr Robert Suhadolnik, a nucleoside analogue expert at Temple University, led to a job offer. "I thought I'd work there for a year, learn, and then return to Hungary."

But things didn't turn out that way. A complicated parting of the ways with Suhadolnik led to a position at Bethesda and then to work with Elliot Barnathan, a cardiologist at the University of Pennsylvania. "Elliot was working on plasminogen activators, molecules that help to dissolve blood clots and their use in minimising the chances of clots forming in the blood vessels. He was planning to do this using DNA, but I suggested mRNA instead. Even though I hadn't synthesised it yet, the science of mRNA was advancing fast."

A series of setbacks and breakthroughs followed over the next few years. "The setbacks were on full display, though the breakthroughs were hardly mentioned at the time. But at the end we had succeeded in using mRNA to make a specific protein within cells."

And then, at Penn, she met Drew Weissman, who shares the Nobel with her. "He was an immunologist without RNA experience, and I was an RNA scientist who didn't know much about immunology. And we needed to find a way by which mRNA could evade detection by the immune system. Finally, after I'd been working on RNA for thirty years, we did it. We could make mRNA in the lab, deliver it into immune cells without causing inflammation."

The paper on this ground-breaking discovery was submitted to Nature, but rejected within 24 hours as merely an 'incremental contribution.' Submitted to Immunity, it was accepted but didn't create waves, not even ripples. This discovery was met with silence. It took a pandemic for it to receive the acclamation it deserved.

The Covid-19-causing SARS-CoV-2 virus was first sequenced in January 2020, and the first vaccines became publicly available less than a year later. Not only did the mRNA vaccines limit mortality and morbidity to an impressive extent, but the vaccines have opened the way to many other applications of the technology – not just vaccines but many other therapies for acquired and genetic diseases. She will be telling the conference about the decades of scientific achievements that preceded this; from the discovery of mRNA in 1961, then the generation of mRNA in a test tube in 1984 and finally evaluating

it in animals as a vaccine against infectious diseases and cancer. "A great extent of progress toward a viable treatment was made during those years. We warded off the immune response and demonstrated that mRNA formulated with lipid nanoparticles could be a potent vaccine. These discoveries eventually led to the development of the mRNA vaccine that has now helped to fight the global pandemic and opened the door for developing breakthrough therapeutics for incurable diseases and unmet medical needs."

The lesson from all of this, says Katalin Karikó, is don't ever give up. "Trust what's inside you. Nurture what you find there, especially when no-one else does. And be prepared to go out and explain your research, particularly if the science is difficult to understand and risks being misrepresented. If people are to benefit from life-saving medical advances, it is our duty to close the gap between what they know and what they need to know."

Interview with Sylvia Metcalfe, ELPAG Lecturer 2025

by Mary Rice, ESHG Press Officer

Sylvia Metcalfe is a former Professor of Medical Genetics in the Department of Paediatrics at the University of Melbourne, Australia and former Group Leader of Genetics Education and Health Research/Genomics and Society at the Murdoch Children's Research Institute. She will be giving the ELPAG Award Lecture on Saturday 24 May at 14.15hrs.

Sylvia Metcalfe first became interested in science as a teenager at high school in England, where she was born. "I particularly liked biology but my interest grew after reading the science fiction book by John Wyndham called The Trouble with Lichen. The protagonist was a woman biochemist and this led to my reading about biochemistry, and I found it fascinating. Also, my Italian grandfather who was living with us at the time died of oesophageal cancer and that in-

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fluenced my interest in science going forward."

Her high school was an all-girls Catholic establishment and quite progressive for its time, she says. "I was there in the 1960s and 70s and the school was already very committed to teaching science. Even though my family was not at all academic, my parents were delighted that I decided to pursue a career in science, and gave me lots of encouragement."

After leaving school, she undertook a BSc in Clinical Biochemistry at Surrey University followed by a PhD in lab-based toxicology/cancer research, with post-docs in London and in New York. "I came to Melbourne in Australia for personal reasons (my partner was Australian) and continued with lab-based research in different areas but was increasingly involved with teaching. So, I came to genetics relatively late in my career (after a short break having children) to teach medical genetics at The University of Melbourne. It was at this time, around 1997, that I embarked on a completely different research path in social science at the university and the Murdoch Children's Research Institute and never looked back!"

She retired from her university role and research group leadership in 2018, although she has continued with honorary positions and consulting until quite recently and is still publishing. Making the switch from lab-based research to research that involved interacting directly with people was definitely the best career decision she took, she says. "As much as I loved research at the bench, it is researching people that I find more gratifying. And surrounding myself with students, both in teaching and research. While they learn from us as lecturers, supervisors and mentors, it is not a cliché to say that I have learned so much from working with them."

Sylvia Metcalfe played a leading role in setting up the Genioz project, which launched in 2015 and set out to understand Australians' understanding of online genomic testing. Their early findings have informed subsequent stages of the study, and thus contributed to strategies for supporting Australians to understand and make meaningful and well-considered decisions about the benefits, harms, and implications of personal genomic tests. She is concerned, though, that finding funding for such projects has become more challenging, particularly at present when more and more people are attracted to non-scientific health beliefs and treatments.

Since retirement, travelling has been high on her wish list. "I have been lucky to have been able to visit so many new places, and will continue to do so. Travelling has also allowed me to practise speaking other languages, something else I decided that I should try to do in retirement." She is also treasurer of her local rugby club, where her husband is president. "I have been able to bring in some grants to the club. It was interesting to see how skills in raising finance for research projects can be quite transferable to other settings!"

Other interests include keeping chickens ("I enjoy spending zen time with them as well as eating their delicious eggs"), painting and picture-framing. The chickens will have to cope without her while she attends ESHG, though, where she will tell the conference about the three main areas of research she has been involved in over the last 25 years or so. "These areas revolve around people's understanding of genetics, genetics/genomics education, informed decision-making (including population genetic screening) and family communication. I won't have time to go into all of this, but I will do my best to give an overall flavour of the research that led to the ELPAG Award."

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EUROPEAN SOCIETY OF HUMAN GENETICS

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AGENDA
General Assembly 2025
Monday, May 26, 2025, 12:15 - 13:15 hrs
Room Brown 3, Allianz MiCo Milan, Italy

1. Welcome by the President of the Society, William Newman
2. Approval of the Agenda and of the timely invitation of members
3. Approval of the Minutes of the last GA in 2024
4. Report on the Activity of the Society 2024-2025, William Newman
5. Financial Report of the Treasurer 2024, Gunnar Houge
6. Reports of the Auditors 2024, Natalia Oliva Teles, Lars Fredrik Engebretsen
7. Exoneration of the Treasurer
8. Exoneration of the Executive Board
9. Opening by the new President of the Society, Martina Cornel
10. Results of the nomination for President-Elect 2025 – Confirmation vote by the Membership
11. Vote on New Deputy Secretary General
12. Results Board Member nominations 2025 - Confirmation vote by the Membership
13. Thanks to leaving Board Members
14. Vote on Statute Amendments
15. Vote on Membership Fees 2026
16. Update on sites of future European Human Genetics Conferences
17. Any other business